



CRX gene

cone-rod homeobox

Normal Function

The *CRX* gene provides instructions for making a protein called the cone-rod homeobox protein. This protein is found in the eyes, specifically in the light-sensitive tissue at the back of the eye called the retina. The cone-rod homeobox protein attaches (binds) to specific regions of DNA and helps control the activity of particular genes. On the basis of this action, this protein is called a transcription factor.

In the retina, the cone-rod homeobox protein is necessary for the normal development of light-detecting cells called photoreceptors. Through its actions as a transcription factor, the cone-rod homeobox protein helps photoreceptor cells mature into two types: rods and cones. Rods are needed for vision in low light, while cones are needed for vision in bright light, including color vision. The protein also helps maintain these cells and preserve vision.

Health Conditions Related to Genetic Changes

cone-rod dystrophy

More than 20 mutations in the *CRX* gene have been found to cause cone-rod dystrophy. The problems associated with this condition include a loss of visual sharpness (acuity), an increased sensitivity to light (photophobia), and impaired color vision. These vision problems worsen over time. Cone-rod dystrophy is caused by mutations that occur in one of the two copies of the *CRX* gene in each cell. *CRX* gene mutations are responsible for about one-quarter of the cases of a form of the condition called autosomal dominant cone-rod dystrophy. These mutations lead to a reduction in the amount of functional cone-rod homeobox protein that is available to regulate other genes in the retina. As a result, maintenance of the rod and cone cells is insufficient and these cells deteriorate over time, leading to the vision problems characteristic of cone-rod dystrophy. Researchers believe that there is enough cone-rod homeobox protein function to allow for photoreceptor cell differentiation, but long-term maintenance of the cells cannot be sustained.

Several *CRX* gene mutations have been found to cause different forms of vision loss in different individuals. It is unclear how mutations in the *CRX* gene can cause different eye disorders.

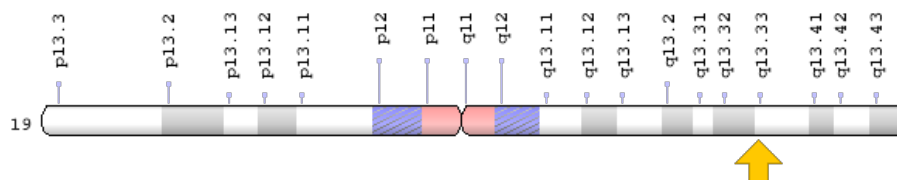
Leber congenital amaurosis

retinitis pigmentosa

Chromosomal Location

Cytogenetic Location: 19q13.33, which is the long (q) arm of chromosome 19 at position 13.33

Molecular Location: base pairs 47,821,842 to 47,843,330 on chromosome 19 (Homo sapiens Annotation Release 108, GRCh38.p7) (NCBI)



Credit: Genome Decoration Page/NCBI

Other Names for This Gene

- cone-rod homeobox protein
- CORD2
- CRD
- LCA7
- orthodenticle homeobox 3
- OTX3

Additional Information & Resources

Educational Resources

- Neuroscience (second edition, 2001): Functional Specialization of the Rod and Cone Systems
<https://www.ncbi.nlm.nih.gov/books/NBK10850/>
- Webvision: The Organization of the Retina and Visual System: Structure of Rods and Cones
<https://www.ncbi.nlm.nih.gov/books/NBK52768/#FuPhototr>
an.2_Structure_of_rods_and_cone

GeneReviews

- Leber Congenital Amaurosis
<https://www.ncbi.nlm.nih.gov/books/NBK1298>
- Nonsyndromic Retinitis Pigmentosa Overview
<https://www.ncbi.nlm.nih.gov/books/NBK1417>

Scientific Articles on PubMed

- PubMed
<https://www.ncbi.nlm.nih.gov/pubmed?term=%28%28CRX%5BTIAB%5D%29+OR+%28cone-rod+homeobox%5BTIAB%5D%29%29+AND+%28%28Genes%5BMH%5D%29+OR+%28Genetic+Phenomena%5BMH%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+1800+days%22%5Bdp%5D>

OMIM

- CONE-ROD HOMEBOX-CONTAINING GENE
<http://omim.org/entry/602225>

Research Resources

- Atlas of Genetics and Cytogenetics in Oncology and Haematology
http://atlasgeneticsoncology.org/Genes/GC_CRX.html
- ClinVar
<https://www.ncbi.nlm.nih.gov/clinvar?term=CRX%5Bgene%5D>
- HGNC Gene Family: PRD class homeoboxes and pseudogenes
<http://www.genenames.org/cgi-bin/genefamilies/set/521>
- HGNC Gene Symbol Report
http://www.genenames.org/cgi-bin/gene_symbol_report?q=data/hgnc_data.php&hgnc_id=2383
- NCBI Gene
<https://www.ncbi.nlm.nih.gov/gene/1406>
- UniProt
<http://www.uniprot.org/uniprot/O43186>

Sources for This Summary

- OMIM: CONE-ROD HOMEBOX-CONTAINING GENE
<http://omim.org/entry/602225>
- Huang L, Li S, Xiao X, Jia X, Wang P, Guo X, Zhang Q. Screening for variants in 20 genes in 130 unrelated patients with cone-rod dystrophy. Mol Med Rep. 2013 Jun;7(6):1779-85. doi: 10.3892/mmr.2013.1415. Epub 2013 Apr 5.
Citation on PubMed: <https://www.ncbi.nlm.nih.gov/pubmed/23563732>

- Huang L, Xiao X, Li S, Jia X, Wang P, Guo X, Zhang Q. CRX variants in cone-rod dystrophy and mutation overview. *Biochem Biophys Res Commun*. 2012 Oct 5;426(4):498-503. doi: 10.1016/j.bbrc.2012.08.110. Epub 2012 Aug 30.
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- Huang L, Zhang Q, Li S, Guan L, Xiao X, Zhang J, Jia X, Sun W, Zhu Z, Gao Y, Yin Y, Wang P, Guo X, Wang J, Zhang Q. Exome sequencing of 47 chinese families with cone-rod dystrophy: mutations in 25 known causative genes. *PLoS One*. 2013 Jun 11;8(6):e65546. doi: 10.1371/journal.pone.0065546. Print 2013.
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<https://ghr.nlm.nih.gov/gene/CRX>

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